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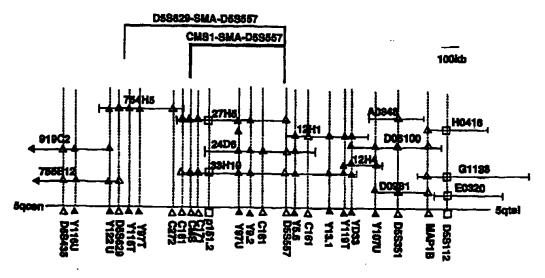
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(54) Title: NEURONAL APOPTOSIS INHIBITOR PROTEIN, GENE SEQUENCE AND MUTATIONS CAUSATIVE OF SPINAL MUSCULAR ATROPHY



(57) Abstract

The gene for the autosomal recessive neurodegenerative disorder Spinal Muscular Atrophy has been mapped to a region of chromosome 5. The gene encodes a protein having homology with apoptosis inhibitor proteins of viruses so that the encoded protein has been labelled as a neuronal apoptosis inhibitor protein (NAIP). A deletion in the (NAIP) domain was identified in persons with Type I, II and III Spinal Muscular Atrophy (SMA) and not in the normal non-SMA population.